European Reference Network
for rare or low prevalence complex diseases

Network Neuromuscular Diseases (ERN EURO-NMD)

Newborn Screening in Neuromuscular Diseases

Satellite Scientific Symposium organized by ERN EURO-NMD
March, 6th 2025

SMA NBS seen by patients and carers

Marie-Christine Ouillade
SMA Europe

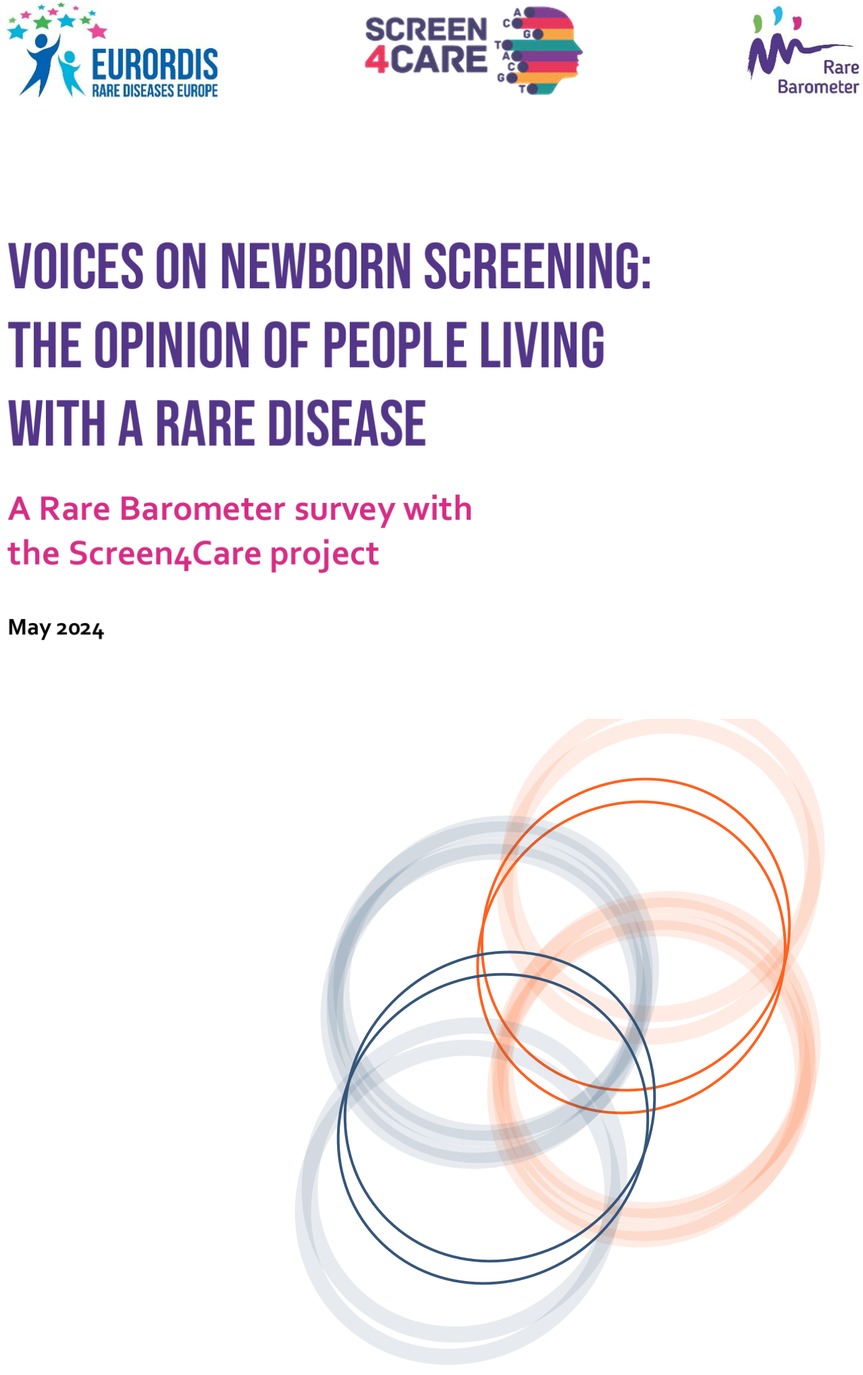
Funded by the European Union

Summary

- Acceptance of SMA NBS
- After a positive screening , the parents decision
- Patient organisation information
- The rôle of SMA Europe

Acceptance of NBS

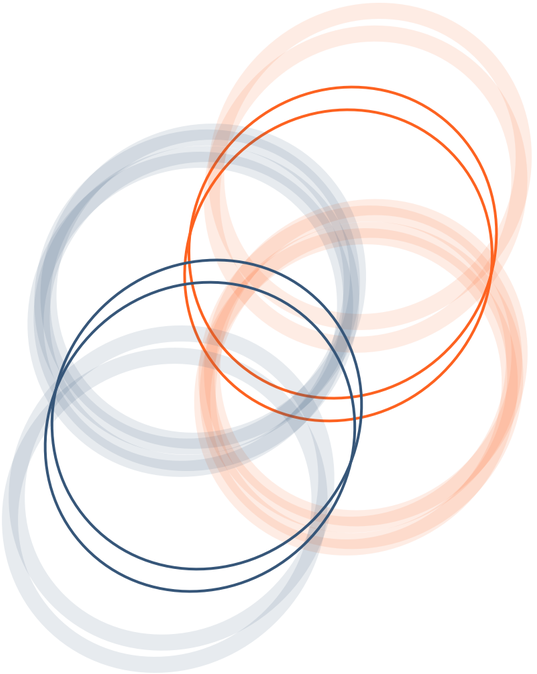
- **How is newborn screening perceived by the rare disease population?**
- Eurordis Rare Barometer survey with the Screen4Care project (Eurordis, 2024)
- 6,179 people living with a rare disease and family members worldwide,
- 5,569 of whom were living in Europe
- 1,300 distinct rare diseases
- “Respondents’ answers confirm the strong support for newborn screening from the rare disease community. They also show that people living with a rare disease and their family members mostly see newborn screening as a way to alleviate the burden of the diagnosis odyssey and to enable parents to make informed choices for their child living with severe and early onset conditions, regardless of their access to a treatment or intervention.”
- (Eurordis, 2024)



Acceptance of SMA NBS

- **How is SMA newborn screening perceived by the public, parents, and adults with SMA?**

- Boardman et al. (Boardman F. K., 2018) online survey to families affected by SMA and the UK public.
- 84%of the public were in favour of introducing SMA NBS,
  - mainly due to the belief that this would result in better health care and life expectancy for the affected infants.
- The majority of SMA adults were also in favour of newborn screening (74%) (Boardman, 2018) as were a mixed population of families and adults (70%), despite preferring pre-conception and / or prenatal screening (Boardman, 2017).



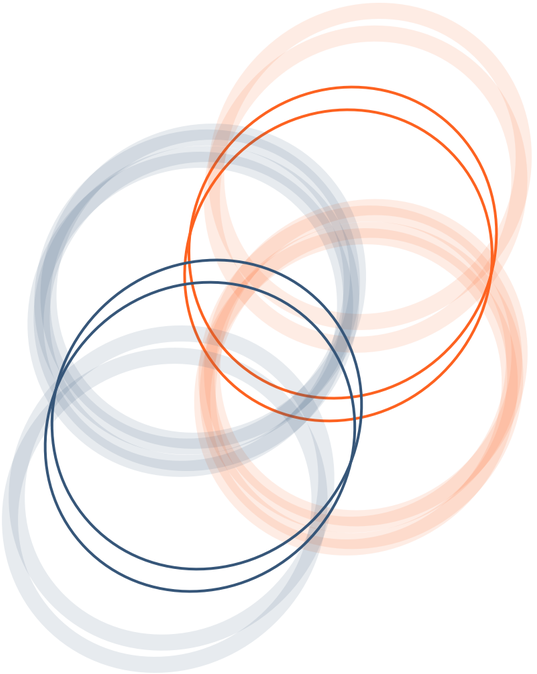
Acceptance of SMA NBS

- **How is SMA newborn screening is accepted by the general population**

- During the different pilots around Europe, the SMA test was proposed to parents with a short explanation of the disease

- In Germany and in France during the pilot and explicit acceptance was requested

- The rate of acceptance of the test was up to 95% by the new parents



After a positive screening

- The need of a quick answer
- The way the information is given to family is crucial

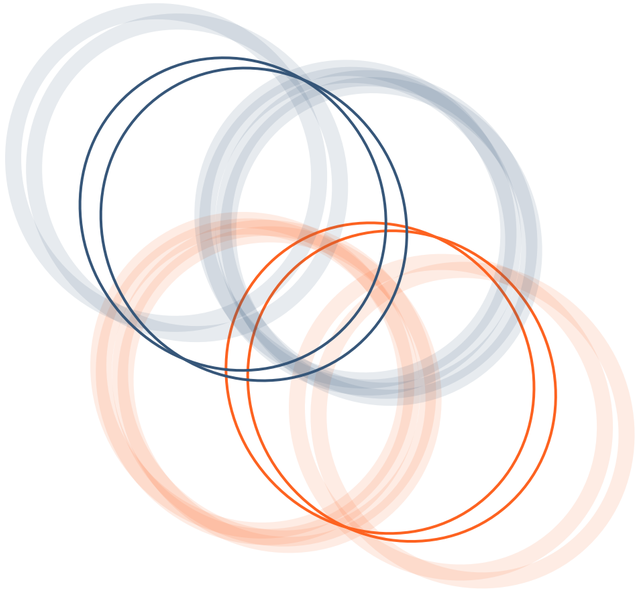
- The timeline is very short between the positive screening and the first treatment

- Around 50% of babies with 2 copies show light symptoms after 3 weeks

- Parents are requested to answer as quickly as possible

Communication

- The family is shocked
  - Less than 20% of the doctor talk is memorised
  - Importance to have a report or a paper after the first information before going back home
  - The annoncement must be absolutly different from the usual way to announce SMA
- Few family doctors are able to give answers to the family
  - Importance of a contact between the center of excellence and the local doctor
- Patient organisation are rarely contacted at this stage
- Very poor information about presymptomatic on internet
  - Risk of misunderstanding



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type of SMA

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Aperçu IA

En savoir plus

Spinal muscular atrophy (SMA) is classified into types 1, 2, 3, and 4 based on the age at which symptoms appear. There are also rarer forms of SMA.

Types of SMA

- **Type 1**: The most severe form of SMA, also known as infantile-onset SMA or Werdnig-Hoffmann disease. Symptoms appear within the first six months of life.
- **Type 3**: A milder form of SMA that develops in children 18 months or older.
- **Type 4**: An adult form of SMA that rarely impacts lifespan.

Other types of SMA

- **Type 0**: Results in death at birth or within one month of life.
- **Spinal and Bulbar Muscular Atrophy (SBMA)**: An adult onset form of SMA, also known as Kennedy's Disease.

SMA characteristics

SMA is an inherited neuromuscular disorder that causes progressive muscle weakness. It's caused by changes to a gene on chromosome 5 called SMN1.

SMA treatment

Noninvasive ventilation and airway clearance techniques are commonly used to improve respiratory insufficiency in people with SMA.

Spinal Muscular Atrophy (SMA) | Boston Children's

Boston Children's Hospital

SMA (Spinal Muscular Atrophy): What It Is, Symptoms & Types

SMA type 0 results in death at birth or within one month of life. SMA type 1 often results in death by the age of 2 without…

Cleveland Clinic

Spinal Muscular Atrophy (SMA) - Johns Hopkins Medicine

Read more. Types of SMA. Spinal muscle atrophy is classified as type 1, 2, 3 or 4 depending on age of onset. Most cases ar…

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presymptomatic sma

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Conseil : Affichez les résultats en **français**. Vous pouvez aussi en savoir plus sur le filtrage par langue.

SMA : le Zolgensma est plus efficace en présymptomatique. Les résultats de l'essai SPR1NT montrent que traiter par le Zolgensma avant l'apparition des symptômes dans la SMA est plus efficace qu'après. De nouvelles preuves en faveur du dépistage néonatal de cette maladie.

18 juil. 2022

AFM Téléthon
https://www.afm-telethon.fr › actualites › sma-le-zolgens...

SMA : le Zolgensma est plus efficace en présymptomatique

À propos des extraits optimisés • Commentaires

National Institutes of Health (NIH) (.gov)
https://pubmed.ncbi.nlm.nih.gov › ... · Traduire cette page

Systematic Review of Presymptomatic Treatment for Spinal ...

de K Cooper · 2024 · Cité 2 fois — This systematic review synthesises findings from prospective studies of **presymptomatic treatment for 5q SMA** published up to December 2023.

Institut de Myologie
https://www.institut-myologie.org › Actualités

Traiter en présymptomatique la SMA avec quatre copies de ...

9 juin 2022 — la frontière entre état **présymptomatique** et symptomatique est ténue, surtout pour des pédiatres moins experts de la pathologie. Quant au choix ...

Zolgensma
https://www.zolgensma.com › pre... · Traduire cette page

Presymptomatic SMA - SPR1NT clinical study results

ZOLGENSMA **is a prescription gene therapy used to treat children less than 2 years old** with spinal muscular atrophy (SMA). ZOLGENSMA is given as a one-time ...

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https://pmc.ncbi.nlm.nih.gov › PM... · Traduire cette page

Systematic Review of Presymptomatic Treatment for Spinal ...

de K Cooper · 2024 · Cité 2 fois — **Spinal muscular atrophy (SMA)** causes the degeneration of motor neurons in the spinal cord. Treatments including nusinersen, risdiplam, and onasemnogene ...

SPINRAZA® (nusinersen)
https://www.spinraza.com › home · Traduire cette page

Presymptomatic SMA Study

Super: In an ongoing supportive study, 25 infants who had not yet shown symptoms of

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Systematic Review of Presymptomatic Treatment for Spinal ...

de K Cooper · 2024 · Cité 2 fois — Three single-arm interventional studies assessed three different **presymptomatic SMA treatments** (nusinersen, onasemnogene abeparvovec, and risdiplam). In the ...

Autres questions

How do you treat presymptomatic SMA?

Treatments including **nusinersen, risdiplam, and onasemnogene abeparvovec** have been shown to be effective in reducing symptoms, with recent studies suggesting greater effectiveness when treatment is initiated in the presymptomatic stage. 14 août 2024

PubMed
https://pubmed.ncbi.nlm.nih.gov › ...

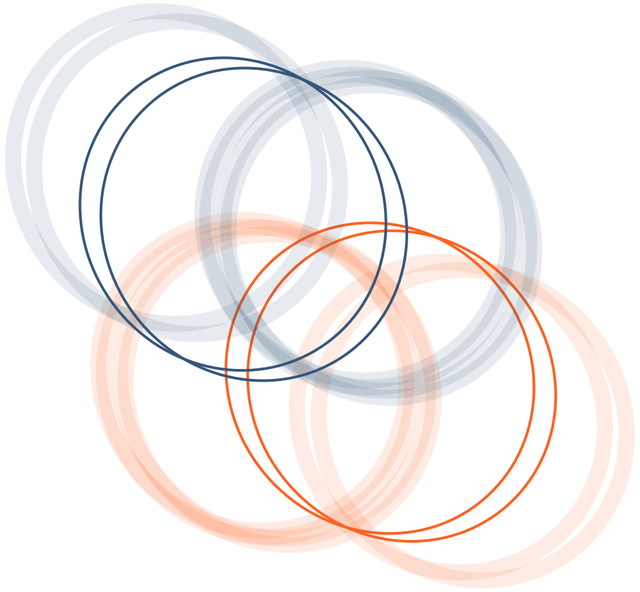
Systematic Review of Presymptomatic Treatment for Spinal Muscular ...

What are the 4 types of SMA?

There are four types of SMA:

The rôle of Patient Organisation in communication

- Very few families contact the PO for a support on decision
- The contact arrives later with questions about the future development of the disaeses
- The Polish information paper

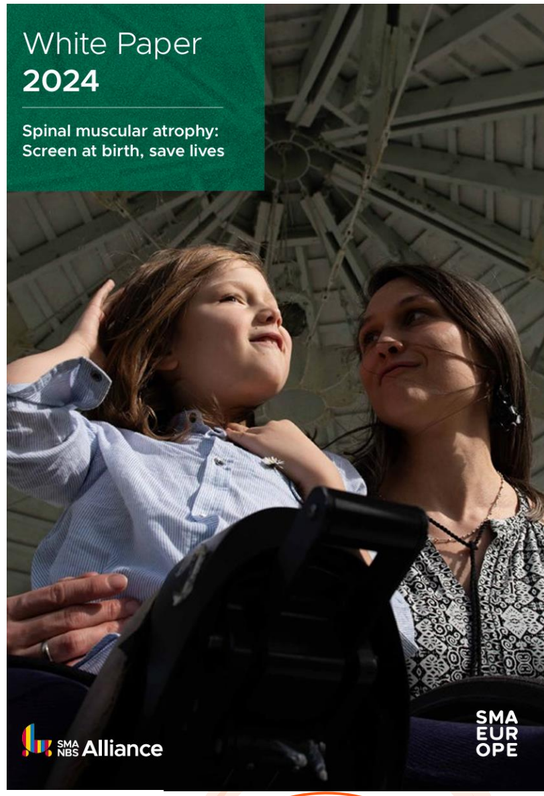


The role of SMA Europe

- **How a patient organisation can accelerate the implementation of SMA NBS**

- July 2020 creation of the NBS SMA Alliance
  - Creation of tools
  - Webinar
  - Scientific conferences
  - The white paper
- End of 2024 72% of the newborns are tested in Europe

- Harmonisation of practices
  - ENMC meeting



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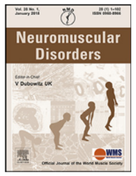
Neuromuscular Disorders

journal homepage: www.elsevier.com/locate/nmd



270th ENMC International Workshop: Consensus for *SMN2* genetic analysis in SMA patients 10–12 March, 2023, Hoofddorp, the Netherlands

Emanuela Abiusi[a,1], Mar Costa-Roger[b,1], Enrico Silvio Bertini[c,2,*], Francesco Danilo Tiziano[a,d,2,*], Eduardo F. Tizzano[b,2,*], on behalf of all participants[3]

[a] *Section of Genomic Medicine, Department of Public Health and Life Sciences, Università Cattolica del Sacro Cuore, Roma, Italy*
[b] *Clinical and Molecular Genetics Area, Vall d'Hebron Hospital; Medicine Genetics Group, Vall d'Hebron Research Institute (VHIR), Barcelona, Spain*
[c] *Research Unit of Neuromuscular Disease, Bambino Gesu' Children's Hospital, IRCCS, Roma, Italy*
[d] *Complex Unit of Medical Genetics, Fondazione Policlinico Universitario IRCCS "A. Gemelli", Roma, Italy*

Thank for your attention

SMA EUROPE

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SMA NBS Alliance